Maple Syrup Urine Disease (MSUD)

Maple Syrup Urine Disease (MSUD) is a very rare inherited autosomal recessive disorder. Newborns with MSUD have a deficiency of the enzyme branched-chain ketoacid dehydrogenase responsible for metabolizing the branched chain amino acids leucine, isoleucine, and valine found in protein. The purpose of newborn screening for MSUD is to identify affected infants rapidly, and initiate treatment to prevent neurological sequelae and death.

Estimated Incidence (MI):	1: 232,000
Laboratory Screening Test:	Leucine using Tandem Mass Spectrometry
Timing of Test:	≥ 24 hours of age: Results are valid
Feeding Effect:	Minimal. Tandem Mass Spectrometry can detect elevations in leucine earlier than previously used qualitative methods.
Transfusion Effect:	None
Confirmation:	All infants with strong and persistent borderline positive tests are referred to the Pediatric Neurology Metabolic Clinic (PNMC) for confirmatory testing and treatment (734) 763-4697.
Treatment:	Immediate treatment with branched-chain free MSUD formula. Specially prepared branched-chain free parenteral nutrition is available for acutely ill infants. Long-term treatment consists of a strict diet limiting the intake of branched chain amino acids while maintaining normal growth and development.

For further information, contact the Newborn Screening Program
Telephone (517) 335-9205
mdch-newbornscreening@michigan.gov

4/2003